ataxia with vitamin E deficiency

Ataxia with vitamin E deficiency is a disorder that impairs the body's ability to use vitamin E obtained from the diet. Vitamin E is an antioxidant, which means that it protects cells in the body from the damaging effects of unstable molecules called free radicals. A shortage (deficiency) of vitamin E can lead to neurological problems, such as difficulty coordinating movements (ataxia) and speech (dysarthria), loss of reflexes in the legs (lower limb areflexia), and a loss of sensation in the extremities (peripheral neuropathy). Some people with this condition have developed an eye disorder called retinitis pigmentosa that causes vision loss. Most people who have ataxia with vitamin E deficiency start to experience problems with movement between the ages of 5 and 15 years. The movement problems tend to worsen with age.

Frequency

Ataxia with vitamin E deficiency is a rare condition; however, its prevalence is unknown.

Genetic Changes

Mutations in the *TTPA* gene cause ataxia with vitamin E deficiency. The *TTPA* gene provides instructions for making the α-tocopherol transfer protein (αTTP), which is found in the liver and brain. This protein controls distribution of vitamin E obtained from the diet (also called α-tocopherol) to cells and tissues throughout the body. Vitamin E helps cells prevent damage that might be done by free radicals.

TTPA gene mutations impair the activity of the αTTP protein, resulting in an inability to retain and use dietary vitamin E. As a result, vitamin E levels in the blood are greatly reduced and free radicals accumulate within cells. Nerve cells (neurons) in the brain and spinal cord (central nervous system) are particularly vulnerable to the damaging effects of free radicals and these cells die off when they are deprived of vitamin E. Nerve cell damage can lead to problems with movement and other features of ataxia with vitamin E deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ataxia with isolated vitamin E deficiency
- AVED
- familial isolated vitamin E deficiency
- FIVE
- Friedreich ataxia phenotype with selective vitamin E deficiency
- Friedreich-like ataxia

Diagnosis & Management

These resources address the diagnosis or management of ataxia with vitamin E deficiency:

- GeneReview: Ataxia with Vitamin E Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1241
- Genetic Testing Registry: Ataxia with vitamin E deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848533/
- MedlinePlus Encyclopedia: Retinitis pigmentosa
  https://medlineplus.gov/ency/article/001029.htm
- MedlinePlus Encyclopedia: Vitamin E
  https://medlineplus.gov/ency/article/002406.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Retinitis pigmentosa
  https://medlineplus.gov/ency/article/001029.htm
- Encyclopedia: Vitamin E
  https://medlineplus.gov/ency/article/002406.htm
- Health Topic: Movement Disorders
  https://medlineplus.gov/movementdisorders.html
- Health Topic: Neuromuscular Disorders
  https://medlineplus.gov/neuromusculardisorders.html

Genetic and Rare Diseases Information Center

- Ataxia with vitamin E deficiency
  https://rarediseases.info.nih.gov/diseases/8595/ataxia-with-vitamin-e-deficiency

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
  https://www.ninds.nih.gov/Disorders/All-Disorders/Ataxias-and-Cerebellar-or-Spinocerebellar-Degeneration-Information-Page
- Office of Dietary Supplements
  https://ods.od.nih.gov/factsheets/VitaminE-Consumer/

Educational Resources

- Disease InfoSearch: Ataxia with Vitamin E Deficiency
  http://www.diseaseinfosearch.org/Ataxia+with+Vitamin+E+Deficiency/639
- MalaCards: ataxia with vitamin e deficiency
  http://www.malacards.org/card/ataxia_with_vitamin_e_deficiency
- Merck Manual Home Edition for Patients and Caregivers: Coordination Disorders
  http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/movement-disorders/coordination-disorders

Patient Support and Advocacy Resources

- Ataxia UK
  https://www.ataxia.org.uk/
- Foundation Fighting Blindness: Retinitis Pigmentosa
  http://www.blindness.org/retinitis-pigmentosa
- National Ataxia Foundation
  http://www.ataxia.org/

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/ataxia-with-vitamin-e-deficiency/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/ataxia.html

GeneReviews

- Ataxia with Vitamin E Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1241

Genetic Testing Registry

- Ataxia with vitamin E deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848533/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22ataxia+with+vitamin+E+deficiency%22+OR+%22Vitamin+E+Deficiency%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ataxia+with+vitamin+E+deficiency%29+OR+%28AVED%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF
  http://omim.org/entry/277460

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